MYO5B gene

myosin VB

Normal Function

The *MYO5B* gene provides instructions for making a protein called myosin Vb. This protein is one of a group of proteins with similar structures called myosins, which are involved in cell movement and the transport of materials within and between cells. Myosin Vb helps to determine the position of various components within cells (cell polarity). Myosin Vb also plays a role in moving components from the cell membrane to the interior of the cell for recycling.

Health Conditions Related to Genetic Changes

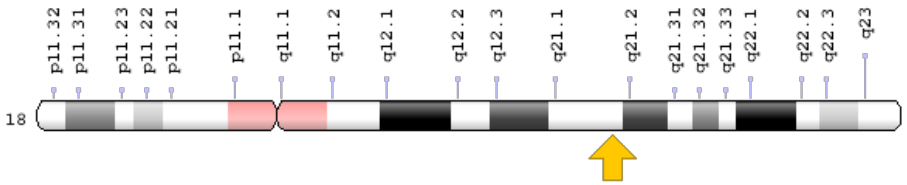
microvillus inclusion disease

More than 40 mutations in the *MYO5B* gene have been found to cause microvillus inclusion disease. This condition is characterized by chronic, life-threatening diarrhea beginning in infancy. The *MYO5B* gene mutations that cause this condition result in a decrease or absence of myosin Vb function. In cells that line the small intestine (enterocytes), a lack of myosin Vb function changes the cell polarity. As a result, enterocytes cannot properly form structures called microvilli, which normally project like small fingers from the surface of the cells and absorb nutrients and fluids from food as it passes through the intestine. Inside affected enterocytes, small clumps of abnormal microvilli mix with misplaced digestive proteins to form microvillus inclusions, which contribute to the dysfunction of enterocytes. Disorganized enterocytes with poorly formed microvilli reduce the intestine's ability to take in nutrients and fluids. The inability to absorb nutrients and fluids during digestion leads to severe diarrhea, malnutrition, and dehydration in individuals with microvillus inclusion disease.

Chromosomal Location

Cytogenetic Location: 18q21.1, which is the long (q) arm of chromosome 18 at position 21.1

Molecular Location: base pairs 49,822,786 to 50,195,081 on chromosome 18 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIAA1119
- MYO5B variant protein
- myosin-Vb
- unconventional myosin-Vb

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2000): Unconventional Myosins https://www.ncbi.nlm.nih.gov/books/NBK9961/#A1804

Scientific Articles on PubMed

- PubMed https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MYO5B%5BTIAB%5D%29+OR+%28myosin+VB%5BTIAB%5D%29%29+OR+%28myosin-Vb%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- MYOSIN VB http://omim.org/entry/606540

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MYO5B.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MYO5B%5Bgene%5D
- HGNC Gene Family: Myosins, class V
  http://www.genenames.org/cgi-bin/genefamilies/set/1100
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7603
- International Microvillus Inclusion Disease Patient Registry
  http://www.mvid-central.org/molgenis.do?__target=main&select=DataExplorerPlugin
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4645
- UniProt
  http://www.uniprot.org/uniprot/Q9ULV0

Sources for This Summary

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